

Robert H Brown

List of Publications by Year in descending order

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172
papers

34,129
citations

10070

75
h-index

5244

171
g-index

186
all docs

186
docs citations

186
times ranked

29450
citing authors

#	ARTICLE	IF	CITATIONS
1	A randomized <scp>placeboâ€controlled</scp> phase 3 study of mesenchymal stem cells induced to secrete high levels of neurotrophic factors in amyotrophic lateral sclerosis. <i>Muscle and Nerve</i> , 2022, 65, 291-302.	1.0	41
2	AAV gene therapy for Tay-Sachs disease. <i>Nature Medicine</i> , 2022, 28, 251-259.	15.2	49
3	Low-level overexpression of wild type TDP-43 causes late-onset, progressive neurodegeneration and paralysis in mice. <i>PLoS ONE</i> , 2022, 17, e0255710.	1.1	20
4	Suppression of mutant C9orf72 expression by a potent mixed backbone antisense oligonucleotide. <i>Nature Medicine</i> , 2022, 28, 117-124.	15.2	72
5	Imaging Net Retrograde Axonal Transport In Vivo: A Physiological Biomarker. <i>Annals of Neurology</i> , 2022, 91, 716-729.	2.8	6
6	Whole-genome sequencing reveals that variants in the Interleukin 18 Receptor Accessory Protein 3â€™UTR protect against ALS. <i>Nature Neuroscience</i> , 2022, 25, 433-445.	7.1	16
7	Excessive release of inorganic polyphosphate by ALS/FTD astrocytes causes non-cell-autonomous toxicity to motoneurons. <i>Neuron</i> , 2022, 110, 1656-1670.e12.	3.8	33
8	Mutation in protein disulfide isomerase A3 causes neurodevelopmental defects by disturbing endoplasmic reticulum proteostasis. <i>EMBO Journal</i> , 2022, 41, e105531.	3.5	11
9	Poly(GR) and poly(GA) in cerebrospinal fluid as potential biomarkers for C9ORF72-ALS/FTD. <i>Nature Communications</i> , 2022, 13, 2799.	5.8	28
10	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. <i>Nature Medicine</i> , 2021, 27, 1197-1204.	15.2	96
11	AAV-Mediated Gene Therapy for Glycosphingolipid Biosynthesis Deficiencies. <i>Trends in Molecular Medicine</i> , 2021, 27, 520-523.	3.5	8
12	Genetic diversity of axon degenerative mechanisms in models of Parkinson's disease. <i>Neurobiology of Disease</i> , 2021, 155, 105368.	2.1	16
13	Genetic susceptibility to hepatocellular carcinoma in chromosome 22q13.31, findings of a genome-wide association study. <i>JGH Open</i> , 2021, 5, 1363-1372.	0.7	9
14	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	9.4	223
15	Intralingual and Intrapleural AAV Gene Therapy Prolongs Survival in a SOD1 ALS Mouse Model. <i>Molecular Therapy - Methods and Clinical Development</i> , 2020, 17, 246-257.	1.8	17
16	Amyotrophic Lateral Sclerosis: Fuel for the Corticofugal Feud. <i>Annals of Neurology</i> , 2020, 88, 682-684.	2.8	2
17	Extracellular microRNAs in human circulation are associated with miRISC complexes that are accessible to anti-AGO2 antibody and can bind target mimic oligonucleotides. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 24213-24223.	3.3	83
18	Immediate and long-term consequences of COVID-19 infections for the development of neurological disease. <i>Alzheimer's Research and Therapy</i> , 2020, 12, 69.	3.0	367

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19	Longitudinal biomarkers in amyotrophic lateral sclerosis. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1103-1116.	1.7	62
20	<i>SOD1</i> Suppression with Adeno-Associated Virus and MicroRNA in Familial ALS. <i>New England Journal of Medicine</i> , 2020, 383, 151-158.	13.9	151
21	Risk factors for hepatocellular carcinoma (HCC) in the northeast of the United States: results of a case-control study. <i>Cancer Causes and Control</i> , 2020, 31, 321-332.	0.8	20
22	Defective daily temperature regulation in a mouse model of amyotrophic lateral sclerosis. <i>Experimental Neurology</i> , 2019, 311, 305-312.	2.0	5
23	Cortical Spreading Depression Denotes Concussion Injury. <i>Journal of Neurotrauma</i> , 2019, 36, 1008-1017.	1.7	34
24	Modulation of actin polymerization affects nucleocytoplasmic transport in multiple forms of amyotrophic lateral sclerosis. <i>Nature Communications</i> , 2019, 10, 3827.	5.8	54
25	Prospective natural history study of <i>C9orf72</i> ALS clinical characteristics and biomarkers. <i>Neurology</i> , 2019, 93, e1605-e1617.	1.5	29
26	Correction of pseudoexon splicing caused by a novel intronic dysferlin mutation. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 642-654.	1.7	20
27	Nucleic Acid Therapeutics for Neurological Diseases. <i>Neurotherapeutics</i> , 2019, 16, 245-247.	2.1	16
28	A new approach for rare variation collapsing on functional protein domains implicates specific genetic regions in ALS. <i>Genome Research</i> , 2019, 29, 809-818.	2.4	21
29	Human genetics and neuropathology suggest a link between miR-218 and amyotrophic lateral sclerosis pathophysiology. <i>Science Translational Medicine</i> , 2019, 11, .	5.8	37
30	NurOwn, phase 2, randomized, clinical trial in patients with ALS. <i>Neurology</i> , 2019, 93, e2294-e2305.	1.5	95
31	Randomized trial of <i>scp</i> -serine in patients with hereditary sensory and autonomic neuropathy type 1. <i>Neurology</i> , 2019, 92, e359-e370.	1.5	83
32	Finding a Treatment for ALS Will Gene Editing Cut It?. <i>New England Journal of Medicine</i> , 2018, 378, 1454-1456.	13.9	11
33	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	3.8	517
34	TDP-43 gains function due to perturbed autoregulation in a <i>Tardbp</i> knock-in mouse model of ALS-FTD. <i>Nature Neuroscience</i> , 2018, 21, 552-563.	7.1	181
35	Genetics of Amyotrophic Lateral Sclerosis. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2018, 8, a024125.	2.9	151
36	Safe and effective superoxide dismutase 1 silencing using artificial microRNA in macaques. <i>Science Translational Medicine</i> , 2018, 10, .	5.8	59

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37	ALS-associated missense and nonsense TBK1 mutations can both cause loss of kinase function. <i>Neurobiology of Aging</i> , 2018, 71, 266.e1-266.e10.	1.5	59
38	Endoplasmic reticulum stress leads to accumulation of wild-type SOD1 aggregates associated with sporadic amyotrophic lateral sclerosis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 8209-8214.	3.3	88
39	Loss of Sarm1 does not suppress motor neuron degeneration in the SOD1G93A mouse model of amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2018, 27, 3761-3771.	1.4	45
40	Impact of Intralingual AAVrh10ΔmiRSOD1 Injection on Respiratory Function in SOD1 G93A Mice. <i>FASEB Journal</i> , 2018, 32, 743.7.	0.2	0
41	Mutant Profilin1 transgenic mice recapitulate cardinal features of motor neuron disease. <i>Human Molecular Genetics</i> , 2017, 26, ddw429.	1.4	67
42	Further analysis of KIFAP3 gene in ALS patients from Switzerland and Sweden. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2017, 18, 302-304.	1.1	4
43	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	129
44	Poly(GP) proteins are a useful pharmacodynamic marker for <i>C9ORF72</i> -associated amyotrophic lateral sclerosis. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	179
45	Amyotrophic Lateral Sclerosis. <i>New England Journal of Medicine</i> , 2017, 377, 1602-1602.	13.9	118
46	Amyotrophic Lateral Sclerosis. <i>New England Journal of Medicine</i> , 2017, 377, 162-172.	13.9	1,264
47	A C9ORF72 BAC mouse model recapitulates key epigenetic perturbations of ALS/FTD. <i>Molecular Neurodegeneration</i> , 2017, 12, 46.	4.4	22
48	Silencing strategies for therapy of SOD1-mediated ALS. <i>Neuroscience Letters</i> , 2017, 636, 32-39.	1.0	55
49	<i>ALS</i> -linked protein disulfide isomerase variants cause motor dysfunction. <i>EMBO Journal</i> , 2016, 35, 845-865.	3.5	109
50	Attenuated traumatic axonal injury and improved functional outcome after traumatic brain injury in mice lacking <i>Sarm1</i> . <i>Brain</i> , 2016, 139, 1094-1105.	3.7	155
51	Mutant SOD1 protein increases Nav1.3 channel excitability. <i>Journal of Biological Physics</i> , 2016, 42, 351-370.	0.7	8
52	Mutant PFN1 causes ALS phenotypes and progressive motor neuron degeneration in mice by a gain of toxicity. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E6209-E6218.	3.3	85
53	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1043-1048.	9.4	494
54	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	9.4	218

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55	Decoding ALS: from genes to mechanism. <i>Nature</i> , 2016, 539, 197-206.	13.7	1,533
56	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , 2016, 7, 11253.	5.8	174
57	Adeno-associated virus delivered artificial microRNA extends survival and delays paralysis in an amyotrophic lateral sclerosis mouse model. <i>Annals of Neurology</i> , 2016, 79, 687-700.	2.8	78
58	Association of a Locus in the <i>CAMTA1</i> Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2016, 73, 812.	4.5	57
59	Therapeutic rAAVrh10 Mediated <i>SOD1</i> Silencing in Adult <i>SOD1</i> ^{G93A} Mice and Nonhuman Primates. <i>Human Gene Therapy</i> , 2016, 27, 19-31.	1.4	85
60	Cross-sectional associations of pulmonary function with systemic inflammation and oxidative stress in individuals with chronic spinal cord injury. <i>Journal of Spinal Cord Medicine</i> , 2016, 39, 344-352.	0.7	20
61	A randomized trial of mexiletine in ALS. <i>Neurology</i> , 2016, 86, 1474-1481.	1.5	72
62	Reply to comment on: A novel dysferlin mutant pseudoexon bypassed with antisense oligonucleotides. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 785-785.	1.7	0
63	International engagement by United States academic neurology departments: A national survey. <i>Annals of Neurology</i> , 2015, 78, 150-152.	2.8	3
64	Amyotrophic Lateral Sclerosis: Review. <i>Seminars in Neurology</i> , 2015, 35, 469-476.	0.5	56
65	The distinct genetic pattern of ALS in Turkey and novel mutations. <i>Neurobiology of Aging</i> , 2015, 36, 1764.e9-1764.e18.	1.5	78
66	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. <i>Science</i> , 2015, 347, 1436-1441.	6.0	823
67	Age-Dependent TDP-43-Mediated Motor Neuron Degeneration Requires GSK3, hat-trick, and xmas-2. <i>Current Biology</i> , 2015, 25, 2130-2136.	1.8	71
68	Juvenile Amyotrophic Lateral Sclerosis. , 2015, , 146-159.		0
69	Identification of rare protein disulfide isomerase gene variants in amyotrophic lateral sclerosis patients. <i>Gene</i> , 2015, 566, 158-165.	1.0	70
70	Emerging mechanisms of molecular pathology in ALS. <i>Journal of Clinical Investigation</i> , 2015, 125, 1767-1779.	3.9	244
71	Endogenous retroviruses in ALS: A reawakening?. <i>Science Translational Medicine</i> , 2015, 7, 307fs40.	5.8	8
72	Human C9ORF72 Hexanucleotide Expansion Reproduces RNA Foci and Dipeptide Repeat Proteins but Not Neurodegeneration in BAC Transgenic Mice. <i>Neuron</i> , 2015, 88, 902-909.	3.8	219

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73	Novel mutations support a role for Profilin 1 in the pathogenesis of ALS. <i>Neurobiology of Aging</i> , 2015, 36, 1602.e17-1602.e27.	1.5	87
74	A novel dysferlin mutant pseudoexon bypassed with antisense oligonucleotides. <i>Annals of Clinical and Translational Neurology</i> , 2014, 1, 703-720.	1.7	24
75	Widespread spinal cord transduction by intrathecal injection of rAAV delivers efficacious RNAi therapy for amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2014, 23, 668-681.	1.4	81
76	Intrinsic Membrane Hyperexcitability of Amyotrophic Lateral Sclerosis Patient-Derived Motor Neurons. <i>Cell Reports</i> , 2014, 7, 1-11.	2.9	583
77	Pathways Disrupted in Human ALS Motor Neurons Identified through Genetic Correction of Mutant SOD1. <i>Cell Stem Cell</i> , 2014, 14, 781-795.	5.2	392
78	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. <i>Neuron</i> , 2014, 84, 324-331.	3.8	308
79	Discovery of a Biomarker and Lead Small Molecules to Target r(GGGGCC)-Associated Defects in c9FTD/ALS. <i>Neuron</i> , 2014, 83, 1043-1050.	3.8	289
80	<scp><i>C9orf72</i></scp> and <scp><i>UNC13A</i></scp> are shared risk loci for amyotrophic lateral sclerosis and frontotemporal dementia: A genome-wide meta-analysis. <i>Annals of Neurology</i> , 2014, 76, 120-133.	2.8	91
81	Mutant SOD1-expressing astrocytes release toxic factors that trigger motoneuron death by inducing hyperexcitability. <i>Journal of Neurophysiology</i> , 2013, 109, 2803-2814.	0.9	99
82	Amyotrophic lateral sclerosis: Problems and prospects. <i>Annals of Neurology</i> , 2013, 74, 309-316.	2.8	117
83	Age of onset of amyotrophic lateral sclerosis is modulated by a locus on 1p34.1. <i>Neurobiology of Aging</i> , 2013, 34, 357.e7-357.e19.	1.5	69
84	Genetic ablation of phospholipase C delta 1 increases survival in SOD1G93A mice. <i>Neurobiology of Disease</i> , 2013, 60, 11-17.	2.1	18
85	The C9ORF72 expansion mutation is a common cause of ALS+FTD in Europe and has a single founder. <i>European Journal of Human Genetics</i> , 2013, 21, 102-108.	1.4	201
86	Mitochondrial damage revealed by immunoselection for ALS-linked misfolded SOD1. <i>Human Molecular Genetics</i> , 2013, 22, 3947-3959.	1.4	78
87	Inhibition of Fast Axonal Transport by Pathogenic SOD1 Involves Activation of p38 MAP Kinase. <i>PLoS ONE</i> , 2013, 8, e65235.	1.1	100
88	Prevalence of Huntington's disease gene CAG repeat alleles in sporadic amyotrophic lateral sclerosis patients. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 265-269.	2.3	15
89	Mutations in the profilin 1 gene cause familial amyotrophic lateral sclerosis. <i>Nature</i> , 2012, 488, 499-503.	13.7	522
90	Association of UBQLN1 mutation with Brown-Vialetto-Van Laere syndrome but not typical ALS. <i>Neurobiology of Disease</i> , 2012, 48, 391-398.	2.1	20

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91	dSarm/Sarm1 Is Required for Activation of an Injury-Induced Axon Death Pathway. <i>Science</i> , 2012, 337, 481-484.	6.0	558
92	EPHA4 is a disease modifier of amyotrophic lateral sclerosis in animal models and in humans. <i>Nature Medicine</i> , 2012, 18, 1418-1422.	15.2	269
93	Mapping of Gene Expression Reveals CYP27A1 as a Susceptibility Gene for Sporadic ALS. <i>PLoS ONE</i> , 2012, 7, e35333.	1.1	50
94	Homologous Recombination Mediates Functional Recovery of Dysferlin Deficiency following AAV5 Gene Transfer. <i>PLoS ONE</i> , 2012, 7, e39233.	1.1	64
95	Mutations in the N-terminal Actin-Binding Domain of Filamin C Cause a Distal Myopathy. <i>American Journal of Human Genetics</i> , 2011, 88, 729-740.	2.6	124
96	Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2011, 70, 964-973.	2.8	168
97	A Yeast Model of FUS/TLS-Dependent Cytotoxicity. <i>PLoS Biology</i> , 2011, 9, e1001052.	2.6	191
98	Dysferlin overexpression in skeletal muscle produces a progressive myopathy. <i>Annals of Neurology</i> , 2010, 67, 384-393.	2.8	33
99	Chromosome 9p21 in sporadic amyotrophic lateral sclerosis in the UK and seven other countries: a genome-wide association study. <i>Lancet Neurology</i> , The, 2010, 9, 986-994.	4.9	205
100	Wild-type and mutant SOD1 share an aberrant conformation and a common pathogenic pathway in ALS. <i>Nature Neuroscience</i> , 2010, 13, 1396-1403.	7.1	600
101	Mutant FUS proteins that cause amyotrophic lateral sclerosis incorporate into stress granules. <i>Human Molecular Genetics</i> , 2010, 19, 4160-4175.	1.4	447
102	Vanadium, aluminum, magnesium and manganese are not elevated in hair samples in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2010, 11, 492-493.	2.3	5
103	miRNA malfunction causes spinal motor neuron disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 13111-13116.	3.3	299
104	Variants of the elongator protein 3 (ELP3) gene are associated with motor neuron degeneration. <i>Human Molecular Genetics</i> , 2009, 18, 472-481.	1.4	512
105	XBP-1 deficiency in the nervous system protects against amyotrophic lateral sclerosis by increasing autophagy. <i>Genes and Development</i> , 2009, 23, 2294-2306.	2.7	463
106	Phase 2 study of sodium phenylbutyrate in ALS. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2009, 10, 99-106.	2.3	135
107	Reduced expression of the <i>Kinesin-Associated Protein 3</i> (<i>KIFAP3</i>) gene increases survival in sporadic amyotrophic lateral sclerosis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 9004-9009.	3.3	177
108	Genome-wide association study identifies 19p13.3 (UNC13A) and 9p21.2 as susceptibility loci for sporadic amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2009, 41, 1083-1087.	9.4	344

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109	DNA sequence analysis of the conserved region around the SOD1 gene locus in recessively inherited ALS. <i>Neuroscience Letters</i> , 2009, 463, 64-69.	1.0	6
110	A Reinnervating MicroRNA. <i>Science</i> , 2009, 326, 1494-1495.	6.0	18
111	The Role of Copy Number Variation in Susceptibility to Amyotrophic Lateral Sclerosis: Genome-Wide Association Study and Comparison with Published Loci. <i>PLoS ONE</i> , 2009, 4, e8175.	1.1	39
112	Evaluation of the Golgi trafficking protein VPS54 (<i>wobbler</i>) as a candidate for ALS. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2008, 9, 141-148.	2.3	31
113	Neuron Research Leaps Ahead. <i>Science</i> , 2008, 321, 1169-1170.	6.0	5
114	A common haplotype within the PON1 promoter region is associated with sporadic ALS. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2008, 9, 306-314.	2.3	37
115	Neonatal Neuronal Circuitry Shows Hyperexcitable Disturbance in a Mouse Model of the Adult-Onset Neurodegenerative Disease Amyotrophic Lateral Sclerosis. <i>Journal of Neuroscience</i> , 2008, 28, 10864-10874.	1.7	219
116	Medications and laboratory parameters as prognostic factors in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2008, 9, 369-374.	2.3	20
117	50bp deletion in the promoter for superoxide dismutase 1 (SOD1) reduces SOD1 expression in vitro and may correlate with increased age of onset of sporadic amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2008, 9, 229-237.	2.3	29
118	Analysis of a genetic defect in the TATA box of the <i>SOD1</i> gene in a patient with familial amyotrophic lateral sclerosis. <i>Muscle and Nerve</i> , 2007, 36, 704-707.	1.0	22
119	Genetic ablation of NMDA receptor subunit NR3B in mouse reveals motoneuronal and nonmotoneuronal phenotypes. <i>European Journal of Neuroscience</i> , 2007, 26, 1407-1420.	1.2	40
120	Dysferlin in Membrane Trafficking and Patch Repair. <i>Traffic</i> , 2007, 8, 785-794.	1.3	138
121	Amyotrophic Lateral Sclerosis: An Emerging Era of Collaborative Gene Discovery. <i>PLoS ONE</i> , 2007, 2, e1254.	1.1	13
122	Variants in candidate ALS modifier genes linked to Cu/Zn superoxide dismutase do not explain divergent survival phenotypes. <i>Neuroscience Letters</i> , 2006, 392, 52-57.	1.0	17
123	Sodium phenylbutyrate prolongs survival and regulates expression of anti-apoptotic genes in transgenic amyotrophic lateral sclerosis mice. <i>Journal of Neurochemistry</i> , 2006, 96, 908-908.	2.1	2
124	ANG mutations segregate with familial and 'sporadic' amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2006, 38, 411-413.	9.4	617
125	Molecular biology of amyotrophic lateral sclerosis: insights from genetics. <i>Nature Reviews Neuroscience</i> , 2006, 7, 710-723.	4.9	984
126	Calpainopathy and eosinophilic myositis. <i>Annals of Neurology</i> , 2006, 59, 875-877.	2.8	33

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127	Case 35-2006. <i>New England Journal of Medicine</i> , 2006, 355, 2132-2142.	13.9	10
128	No association of DYNC1H1 with sporadic ALS in a case-control study of a northern European derived population: A tagging SNP approach. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2006, 7, 46-56.	2.3	9
129	Respiratory dysfunction and management in spinal cord injury. <i>Respiratory Care</i> , 2006, 51, 853-68;discussion 869-70.	0.8	240
130	Sodium phenylbutyrate prolongs survival and regulates expression of anti-apoptotic genes in transgenic amyotrophic lateral sclerosis mice. <i>Journal of Neurochemistry</i> , 2005, 93, 1087-1098.	2.1	315
131	Tetanus toxin fragment C fusion facilitates protein delivery to CNS neurons from cerebrospinal fluid in mice. <i>Journal of Neurochemistry</i> , 2005, 95, 1118-1131.	2.1	23
132	Proteomic profiling of cerebrospinal fluid identifies biomarkers for amyotrophic lateral sclerosis. <i>Journal of Neurochemistry</i> , 2005, 95, 1461-1471.	2.1	191
133	The genetics of amyotrophic lateral sclerosis. , 2005, , 758-771.		0
134	Amyotrophic Lateral Sclerosis – A New Role for Old Drugs. <i>New England Journal of Medicine</i> , 2005, 352, 1376-1378.	13.9	8
135	Disruption of muscle membrane and phenotype divergence in two novel mouse models of dysferlin deficiency. <i>Human Molecular Genetics</i> , 2004, 13, 1999-2010.	1.4	166
136	Mutant dynactin in motor neuron disease. <i>Nature Genetics</i> , 2003, 33, 455-456.	9.4	884
137	Sixteen novel mutations in the Cu/Zn superoxide dismutase gene in amyotrophic lateral sclerosis: a decade of discoveries, defects and disputes. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases</i> , 2003, 4, 62-73.	1.4	234
138	Dysferlin Interacts with Annexins A1 and A2 and Mediates Sarcolemmal Wound-healing. <i>Journal of Biological Chemistry</i> , 2003, 278, 50466-50473.	1.6	336
139	Binding the Abdomen Can Improve Speech in Men With Phrenic Nerve Pacers. <i>American Journal of Speech-Language Pathology</i> , 2002, 11, 71-76.	0.9	7
140	A novel, blood-based diagnostic assay for limb girdle muscular dystrophy 2B and miyoshi myopathy. <i>Annals of Neurology</i> , 2002, 51, 129-133.	2.8	98
141	A gene encoding a putative GTPase regulator is mutated in familial amyotrophic lateral sclerosis 2. <i>Nature Genetics</i> , 2001, 29, 166-173.	9.4	635
142	SPTLC1 is mutated in hereditary sensory neuropathy, type 1. <i>Nature Genetics</i> , 2001, 27, 261-262.	9.4	284
143	SOD1 mutants linked to amyotrophic lateral sclerosis selectively inactivate a glial glutamate transporter. <i>Nature Neuroscience</i> , 1999, 2, 427-433.	7.1	282
144	Molecular genetic analysis of dysferlin in Japanese patients with Miyoshi myopathy. <i>Proceedings of the Japan Academy Series B: Physical and Biological Sciences</i> , 1999, 75, 207-212.	1.6	12

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145	Breathlessness and Exercise in Spinal Cord Injury. <i>Journal of Spinal Cord Medicine</i> , 1999, 22, 297-302.	0.7	13
146	Breathlessness in Spinal Cord Injury Depends on Injury Level. <i>Journal of Spinal Cord Medicine</i> , 1999, 22, 97-101.	0.7	22
147	Dysferlin, a novel skeletal muscle gene, is mutated in Miyoshi myopathy and limb girdle muscular dystrophy. <i>Nature Genetics</i> , 1998, 20, 31-36.	9.4	857
148	SOD1 aggregates in ALS: Cause, correlate or consequence?. <i>Nature Medicine</i> , 1998, 4, 1362-1364.	15.2	54
149	Mutations in the glutamate transporter EAAT2 gene do not cause abnormal EAAT2 transcripts in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 1998, 43, 645-653.	2.8	109
150	Self-Control and External Control of Mechanical Ventilation Give Equal Air Hunger Relief. <i>American Journal of Respiratory and Critical Care Medicine</i> , 1998, 157, 415-420.	2.5	37
151	Inhibition of ICE slows ALS in mice. <i>Nature</i> , 1997, 388, 31-31.	13.7	298
152	Evidence of Increased Oxidative Damage in Both Sporadic and Familial Amyotrophic Lateral Sclerosis. <i>Journal of Neurochemistry</i> , 1997, 69, 2064-2074.	2.1	671
153	An update on superoxide dismutase 1 in familial amyotrophic lateral sclerosis. <i>Journal of the Neurological Sciences</i> , 1996, 139, 10-15.	0.3	20
154	The Dawning of a New Era in the Molecular Biology of the Muscular Dystrophies. <i>Brain Pathology</i> , 1996, 6, 17-17.	2.1	3
155	Dystrophin-Associated Proteins and the Muscular Dystrophies: A Glossary. <i>Brain Pathology</i> , 1996, 6, 19-24.	2.1	46
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157	Sequence variants in human neurofilament proteins: Absence of linkage to familial amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 1996, 40, 603-610.	2.8	78
158	Motor neurons in Cu/Zn superoxide dismutase-deficient mice develop normally but exhibit enhanced cell death after axonal injury. <i>Nature Genetics</i> , 1996, 13, 43-47.	9.4	1,153
159	Apolipoprotein E ϵ 4 allele is not associated with earlier age at onset in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 1995, 38, 460-463.	2.8	80
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161	Superoxide Dismutase Concentration and Activity in Familial Amyotrophic Lateral Sclerosis. <i>Journal of Neurochemistry</i> , 1995, 64, 2366-2369.	2.1	101
162	Linkage of recessive familial amyotrophic lateral sclerosis to chromosome 2q33-q35. <i>Nature Genetics</i> , 1994, 7, 425-428.	9.4	221

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164	Mutations in Cu/Zn superoxide dismutase gene are associated with familial amyotrophic lateral sclerosis. <i>Nature</i> , 1993, 362, 59-62.	13.7	6,331
165	A Met-to-Val mutation in the skeletal muscle Na ⁺ channel β -subunit in hyperkalaemic periodic paralysis. <i>Nature</i> , 1991, 354, 387-389.	13.7	356
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